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LETTERS

THE 100 000 GENOMES PROJECT

The 100 000 Genomes Project: feeding back to patients

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The 100 000 Genomes Project described by Clare Turnbull and colleagues[1] is a world leading initiative offering welcome opportunities for patients with rare and undiagnosed conditions. But the challenges of delivering results cannot be minimised.

Recruitment is behind schedule and only reached halfway in February, three years into the project, which is due to end this October. Reporting times remain longer than planned: we have received results for only a fraction of the 80 plus families recruited in our paediatric dermatology department. The use of targeted gene panels is speeding up turnaround and (helpfully) reducing the number of variants of unknown significance, but this generates many negative reports—patients will not be satisfied with "no mutation found," and those cases require a robust system of further interrogation.

Systems for multidisciplinary discussion and feedback to patients, particularly those recruited outside clinical genetics departments, are not yet established. With colleagues at the West Midlands Genomic Medicine Centre we are building a specialty network to support interpretation of results and incorporation of genomic testing into routine clinical practice after the 100 000 Genomes Project. Our recent regional survey confirmed that only a handful of dermatologists are actively recruiting, but most want to be involved in discussion of their patients' results. Time pressures preclude regular face-to-face multidisciplinary team meetings, but colleagues would access an online conferencing solution.

We strongly support the 100 000 Genomes Project. At our first "dermatogenetic" multidisciplinary team meeting we discussed an undiagnosed disorder with an unexpected mutation, raising the important possibility of a novel disease with an unsuspected susceptibility to cancer. We look forward to seeing the ideals of the 100 000 Genomes Project effectively realised and translated into future NHS genomic testing, but pragmatic solutions must quickly be found to ensure that patients receive validated and useful results in a timely manner.

Competing interests: No competing interests

Full response at: https://www.bmj.com/content/361/bmj.k1687/rr.

1 Turnbull C, Scott RH, Thomas E, etal. 100 000 Genomes Project. The 100 000 Genomes Project: bringing whole genome sequencing to the NHS. *BMJ* 2018;361:k1687. 10.1136/bmj.k1687 29691228

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